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Parental Decisions to Terminate/Continue Following Abnormal Cytogenetic Prenatal Diagnosis: "What" Is Still More Important Than "When"

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This study was undertaken to determine if parental decisions to continue or terminate following the diagnosis of a cytogenetic abnormality have changed over the past 8 years at the same center. Parental decisions in 310 prenatal chromosomal abnormalities were stratified by procedure (chorionic villus sampling [CVS] vs. amniocentesis) and the severity of the anomaly (severe vs mild-moderate). Patients with severe anomalies were much more likely to terminate regardless of gestational age. There was a trend (P = .107) toward a lower rate of termination for mild-moderate degrees in the second trimester. There was no change in patient's decisions over time. Patients' decisions about termination are focused on the severity of the disorder and only marginally influenced by when in gestation the decision is made. © 1996 Wiley-Liss, Inc.

KEY WORDS: prenatal diagnosis, chorionic villus sampling, anomalies, abortion, chromosomal abnormalities

INTRODUCTION

Following the prenatal diagnosis of a severe fetal abnormality, public perception suggests that most couples will choose to terminate the pregnancy [Henry, 1992]. However, extensive experience with the difficulties and anguish faced by such couples when confronted with the diagnosis of an abnormality in an otherwise wanted pregnancy is not at all straightforward [Drugan et al., 1990; Rice, 1992; Pryde et al., 1992; Holmes-Seidel et al., 1987; Kenyon et al., 1988; Vincent et al., 1991]. In the 1980s it was anticipated that with the development of first trimester prenatal diagnosis (particularly before the bonding process was accelerated by ultrasound findings), that a greater percentage of couples having an abnormality diagnosed in the first trimester would choose to terminate as compared to those whose diagnosis was made later in pregnancy [Verp et al., 1988]. Such an association was found by Verp et al. [1988] but refuted by Drugan et al. [1990] who studied the decision of our patient population from 1986 to 1988 who had prenatal diagnosis of chromosome abnormality. In 1992 we asked a slightly different question, i.e., what about the nonaneuploid structural abnormalities? Analogously, Pryde et al. [1992] found that in nonaneuploid abnormalities the main predictor of the decision to abort was the severity of the fetal prognosis. The gestational age at diagnosis was not a particularly important factor in the decision to abort or not. When there was an uncertain prognosis, couples usually opted to continue the pregnancy, especially if there were hope of fetal or neonatal correction of the abnormality even if only investigational.

In this study we have considerably expanded our previous work [Drugan et al., 1990], specifically attempting to discern if there has been a change in patient behavior following the diagnosis of an abnormality, and to see if there is an affect of gestational age at time of diagnosis.

MATERIALS AND METHODS

In this study we have considerably expanded on the original paper by Drugan et al.—now looking at patients over an 8-year period, including 15,959 patients who underwent chorionic villus sampling (CVS) or amniocentesis.

The study population was formed from those patients seen for genetic counseling, ultrasound, and invasive procedures at the Reproductive Genetics Program at Hutzel Hospital/Wayne State University. Our patients come from varying socioeconomic states and ethnic

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backgrounds. About 80% are from the suburbs and about 20% from the inner city. About 70% are white and 30% African American. Over the past several years we have seen a trend of patient mix to increasing complexity and a higher proportion of indigent patients.

Patients' decisions to continue or terminate pregnancies were compared in 3 time periods: 1986–1988, 1989–1991, and 1992–April 1994. The first time period was the material used for our first publication [Drugan et al., 1990]. Patients' decisions following the diagnoses of viable pregnancies with cytogenetic abnormalities were divided by severity of diagnosis. Balanced translocations and intrauterine fetal demises were excluded. The "severe" category was defined as autosomal trisomies, unbalanced translocations, and 45,X with ultrasound abnormalities. The "mild to moderate" category included sex chromosome abnormalities, markers, mosaics, and inversions. These categories were defined, somewhat arbitrarily, several years ago for the 1990 Drugan et al. paper. Admittedly, there have been some modifications about counseling for certain conditions, e.g., trisomy 18 over the 8-year period. However, for consistency and to keep the data sets from being too small to be meaningful, the original definitions have been maintained.

In general "serious" conditions were ones with certain significant expectations of morbidity or mortality. Thus, for example, 45,X with ultrasound abnormalities fits the serious group, but 45,X without does not. Patient counseling for all abnormalities is conducted by the reproductive geneticists (all Ob/Gyn trained as well) and genetic counselors. Virtually all patients and families were counseled by more than one individual with a conscious effort toward nondirectiveness. It is not possible to stratify the data retrospectively by the individual physicians or counselors involved. While there is certainly some individual variations among the specific approaches of the health professionals, overall there is reasonable consistency among the group. The categories "severe" and "mild/moderate" are for the purpose of the study and not regularly presented to patients as such.

Most CVS procedures were performed between 9–12 weeks and most amniocenteses between 14–20 weeks. Terminations were done up to 24 weeks and were done by dilation and curettage (D&C), dilatation and extraction (D&E), or prostaglandin induction as appropriate, with patient wishes in mind. Statistical analysis was by χ^2 as appropriate.

RESULTS

Three hundred ten chromosome abnormalities were detected prior to 24 weeks of gestation, thereby permitting the patients the legal option to consider termination in the state of Michigan. Cytogenetic abnormalities were found in 118 of 5,817 CVSs and 192 of 10,142 amniocenteses. Following a "severe" vs. "mild/moderate" diagnosis, patients were much more likely to terminate for a "severe" diagnosis regardless of the gestational age ($\chi^2 = 108$, $P < .0001$). Differences for the "severe" category between CVS and amniocentesis terminations were not significant and have not changed over time (Tables I and II). The lower rate of "mild to moderate" amniocentesis patients terminating approached significance ($\chi^2 = 2.6$, $P = .107$). However, the time trend was not significant.

DISCUSSION

The major variation in likelihood to terminate between "severe" and "mild to moderate" abnormalities has not changed over the past 8 years. Over 80% of patients with severe abnormalities have chosen to terminate regardless of "when" in gestation the diagnosis was made. The average termination rate for CVS patients with "mild to moderate" disorders was 37% vs. 20% for those on amniocentesis, a difference which approached but did not reach significance. There was a slight trend in the "mild to moderate" group toward a lower rate of pregnancy termination over the past several years, although at this time the trend was insignificant with these numbers.

Despite the vastly increased utilization of ultrasound study in the intervening decade since our first data set, patients' decisions about continuation or abortion of a pregnancy with an abnormality have not changed. We speculate that as patients have become more used to seeing ultrasound figures early in gestation that the emotional impact of the bonding from such is perhaps not as great as it was in the early 1980s [Cox et al., 1987; Fletcher and Evans, 1983]. Therefore, there may be a somewhat less emotional impact of ultrasound in patients dealing with problems as they are diagnosed.

In a pluralistic society, patients confronted with emotionally charged issues such as the diagnosis of a fetus with a significant anomaly can be expected to have differing initial reactions and thoughts about continuation or abortion. Our data suggest that on balance, patients with more severe anomalies have a greater

TABLE I. Patients' Decision to Terminate or Continue Following Abnormalities Discovered on CVS

	CVS										
		Severe					Mild/moderate				
			Terminate		Continue			Terminate		Continue	
	Total	Total	N	%	N	%	Total abnormal	N	%	N	%
1986–1988	1,645	18	16	89	2	11	4	2	50	2	50
1989–1991	2,867	40	35	88	5	12	23	9	39	14	61
1992–4/94	1,305	19	17	89	2	11	14	4	29	10	71
Total 1986–1994	5,817	77	68	88	9	12	41	15	37	26	63

TABLE II. Patients' Decision to Terminate or Continue Following Abnormalities Discovered on Amniocentesis

		Amnio									
		Severe					Mild/moderate				
			Terminate		Continue			Terminate		Continue	
	Total	Total abnormal	N	%	N	%	Total abnormal	N	%	N	%
1986–1988	3,576	40	38	95	2	5	18	4	22	14	78
1989–1991	3,401	36	27	75	9	25	25	6	24	19	76
1992–4/94	3,165	52	42	81	10	19	21	3	14	18	86
Total 1986–1994	10,142	128	107	84	21	16	64	13	20	51	80

likelihood of choosing to terminate. Such data should not be considered surprising. In fact, such data are reassuring of the quality and nondirectiveness of the genetic counseling process that allows couples to reach their own decisions.

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